Supplemental Table. Genetic Characteristics of Affected Patients and Heterozygous Carriers of Choroideremia Study Cohort

Patient	cDNA	Protein	Exon	Type	Coding effect	Align GVGD	SIFT	dbSNP	Clin.sig	Splice effect	MaxEnt	NNSPLICE	SSF
P1	c.877C>T	p.(Arg293*)	Exon 7	Substitution	Nonsense	Class C0 (GV: 241.65 - GD: 77.68)	Deleterious (score: 0)	rs132630266	pathogenic	•			
P2	c.49+1G>A	p.?	Intron 1	Substitution					-	The consequence of this change is not predictable, but a skip of exon 15 is very likely.	-100.00%	-100.00%	-100.00%
Р3	c.1349G>T	p.(Arg450Met)	Exon 10	Substitution	Missense								
P4	c.315-2A>G	p.?	Intron 4	Substitution						The consequence of this change is not predictable, but a skip of exon 5 is very likely.	-100.00%	-100.00%	-100.00%
P5	c.1771-1G>A	p.?	Intron 14	Substitution						The consequence of this change is not predictable, but a skip of exon 15 is very likely.	-100.00%	-100.00%	-100.00%
P6	Deletion of Exon 12												
P7	c.82del	p.(Ser28GInfs*32)	Exon 2	Deletion	Frameshift								
P8	c.1520A>G	p.(His507Arg)	Exon 13	Substitution	Missense	Class C0 (GV: 249.35 - GD: 13.27)	Tolerated (score: 0.16)	rs397514603	pathogenic				
P9	Not screened												
P10	c.1237_1328del	p.(Ser413Valfs*18)	Exon 9	Deletion	Frameshift								
P11	Deletion of Exons 6 and 7												
P12	c.1580_1584del	p.(Leu527Cysfs*5)	Exon 13	Deletion	Frameshift								
P13	Deletion of Exons 9 and 10												
P14	c.1584_1587del	p.(Val529Hisfs*7)	Exon 13	Deletion	Frameshift			rs587776746	pathogenic				
P15	c.819+1G>A	p.?	Intron 6	Substitution						The consequence of this change is not predictable, but a skip of exon 6 is very likely.	-100.00%	-100.00%	-100.00%
P16	Deletion of entire CHM locus												
Carrier													
C1	c.1584_1587del	p.(Val529Hisfs*7)	Exon 13	Deletion	Frameshift			rs587776746	pathogenic				
C2	c.1349G>T	p.(Arg450Met)	Exon 10	Substitution	Missense								
C3	c.49+1G>A	p.?	Intron 1	Substitution						The consequence of this change is not predictable, but a skip of exon 15 is very likely.	-100.00%	-100.00%	-100.00%
C4	Not screened												
C5	Deletion of Exon 12												
C6	c.757C>T	p.(Arg253*)	Exon 6	Substitution	Nonsense			rs886041178	pathogenic				
C7	c.1584_1587del	p.(Val529Hisfs*7)	Exon 13	Deletion	Frameshift			rs587776746	pathogenic				
C8	c.189G>C	p.(Gln63His)	Exon 3	Substitution	Missense	Class C0 (GV: 353.86 - GD: 0.00)	Tolerated (score: 0.19)						
C9	c.940G>A	p.(Gly314Arg)	Exon 7	Substitution	Missense	Class C0 (GV: 236.23 - GD: 41.54)	Tolerated (score: 0.12)						